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Chondrodysplasia Punctata, Humero-Metacarpal Type: A Second Case

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We report on a boy with symmetrical rhizomelic shortness of the upper limbs and punctate epiphyseal calcifications noted at birth. Radiographs documented short and wide humeri, symmetrical brachymetacarpy, coronal clefts of the veretebrae, and punctate calcifications in the spine, sacrum, shoulder, feet, and trachea. Borochowitz [1991] described a similar patient with an apparently new syndrome of chondrodysplasia punctata (CP), distinct from previously described forms. He suggested the term "chondrodysplasia punctata, humero-metacarpal (HM)" type. We present our patient as a second case of this form of CP.

KEY WORDS: chondrodysplasia punctata, punctate epiphyseal calcifications, rhizomelic shortening, short humeri, short metacarpals, skeletal dysplasia

INTRODUCTION

We describe a boy with symmetrical upper limb rhizomelic shortness and chondrodysplasia punctata (CDP). Our patient is very similar to that described by Borochowitz [1991] with an apparently new and distinct form of CDP he called chondrodysplasia punctata, humero-metacarpal (CDP-HM) type. We present this case as a second example of the DP-HM type, confirming this as a unique entity.

CLINICAL REPORT

The boy, 2,110 gm, 43.5 cm long at birth after a 38½-week gestation, was born to a 15-year-old G_1 black woman. The pregnancy was complicated by a gonorrheal infection treated several months before delivery. The mother also smoked one pack per day of cigarettes until the sixth month of gestation. She developed eclampsia and the baby was delivered in emergency by cesarian section. Apgar scores were 9 at 1 and at 5 minutes. The baby was noted to have very short arms with normal legs. Hyperextensibility of his knees resolved by discharge at age 3 days. The family history was unremarkable, and in particular, there were no individuals with short stature or disproportionate skeletal findings.

A skeletal survey obtained on the first day of life documented short humeri (Fig. 1). In addition, stippling was noted (Figs. 2, 3) in the epiphyses of the humeri as well as along the spine, bones of the feet, and left shoulder. There was poor and irregular ossification of the vertebral bodies as well as coronal clefts in the vertebrae. Tracheal ring calcifications were seen as well.

The patient was evaluated by us at age 7 weeks at which time he was noted to have severe bilateral rhizomelic shortness of the arms. Length was 54 cm (10th centile), weight 4.2 kg (25th centile), OFC 42½ cm (10th centile). He was an active boy with a depressed nasal bridge, prominent upper lip, and thick philtrum with a normal palate. Eye findings were normal. The chest appeared somewhat short with a protuberant abdomen without hepatosplenomegaly. There was striking rhizomelic shortness of arms, but the legs were normally proportioned. His hands (Fig. 4) appeared small with a

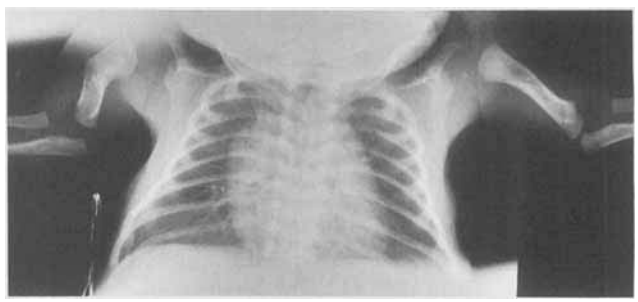

Fig. 1. Roentgenogram of chest and arms at birth with rhizomelic shortness and abnormal appearance of humeri. Note stippling of left humerus.

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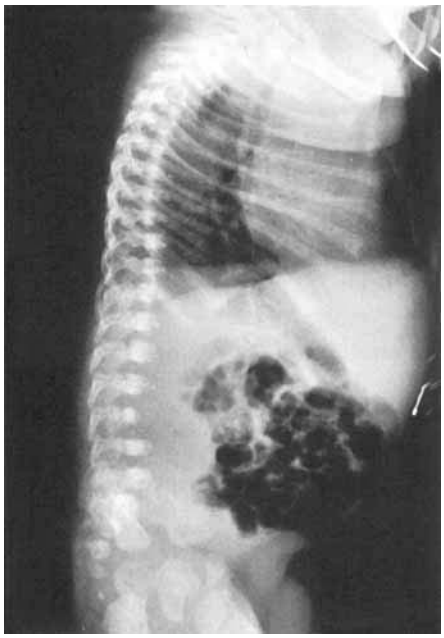

Fig. 2. Lateral roentgenogram of the spine at birth with coronal clefting of the vertebral bodies and stippling along the spinal column.

Fig. 3. Roentgenogram of the legs and feet. Bones of the legs are normal but note stippling of the bones in the left foot.

"trident" configuration. Neurologic status was normal. Prophase chromosomes, very long chain fatty acids, phytanic acid, and plasmalogen levels were normal.

The patient was seen most recently at age 15 months. He has been healthy with normal development. He still has (Fig. 5) striking rhizomelic shortness of arms and

Fig. 4. Roentgenogram of the hands.

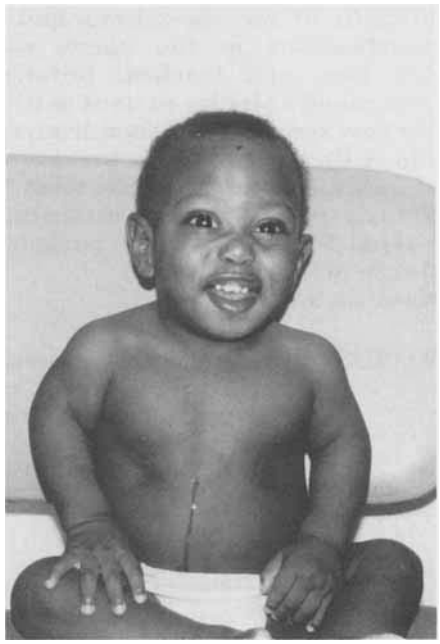

Fig. 5. The patient at age 15 months. Note bilateral rhizomelic shortness of arms, a depressed nasal bridge, prominent upper lip, and thick philtrum.

small hands with a trident configuration. Weight and OFC were at the 10th centile but length was 71 cm, which is below the 5th centile. Length has been below the 5th centile since 6–7 months. His growth chart is shown in Figure 6. Radiographs (Fig. 7) still show stippling in hands and feet, but not in the humeri, along the spine or in the shoulder, as originally seen. Metacarpals were short. There is persistence of coronal clefts, and the humeri continue to show striking rhizomelic shortness.

DISCUSSION

The differential diagnosis of punctate epiphyses in a newborn includes a number of skeletal dysplasias, in-

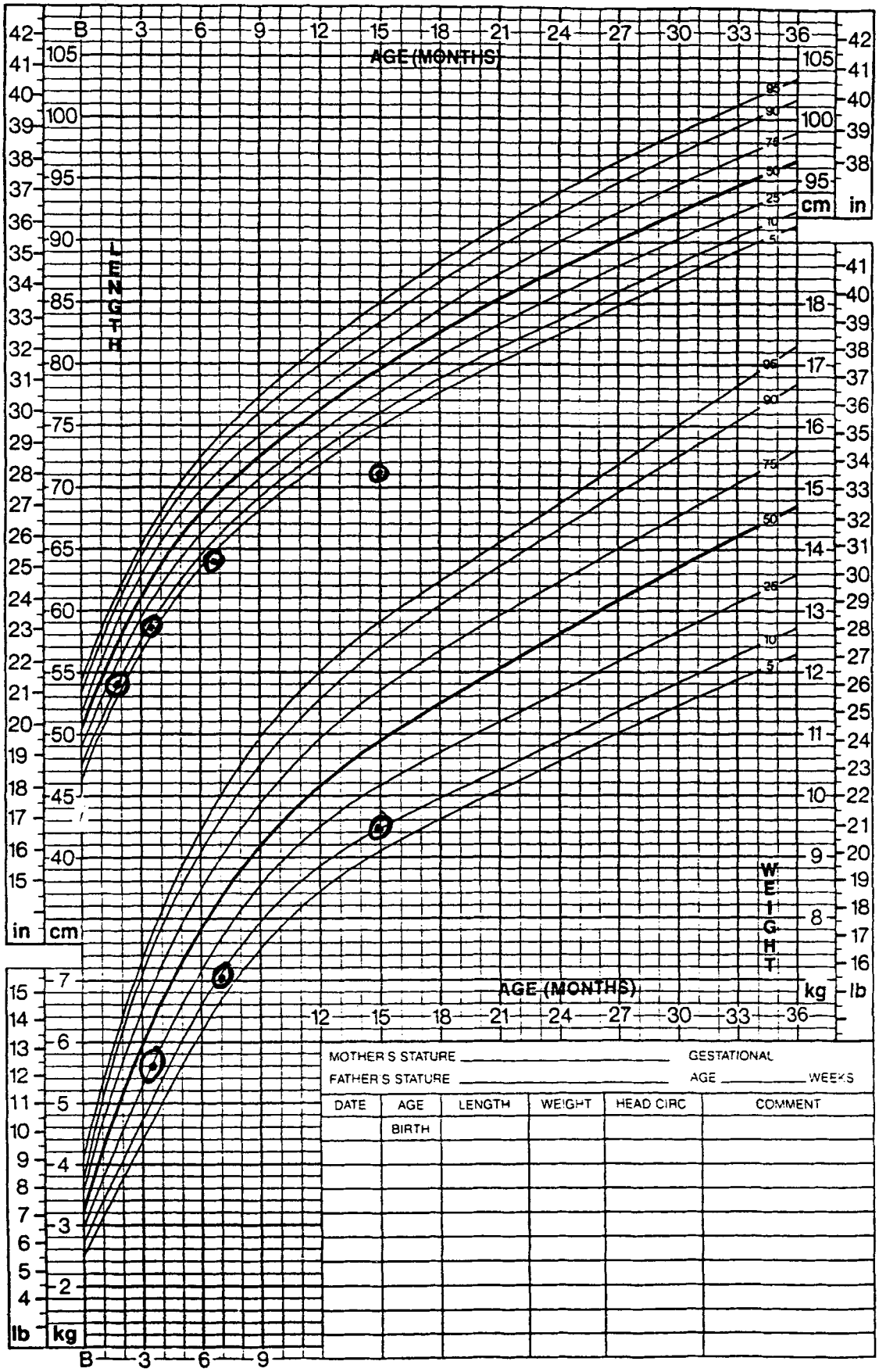

Fig. 6. Growth charts showing deceleration of length growth since age 6–7 months.

Fig. 7. Lateral roentgenogram of the spine at age 15 months showing persistence of coronal clefts but resolution of the stippling along the spine.

born errors of metabolism and storage disorders, chromosomal disorders, and various teratogenic syndromes. Among the skeletal dysplasias, Conradi-Hünerman CDP (an X-linked dominant disorder) [Spranger et al., 1971] and rhizomelic CDP (an autosomal recessive peroxisomal disorder) [Gilbert et al., 1976] are the best known. Poznanski groups several conditions under the term "CDP-metacarpal type" [Poznanski, 1994]. This heterogeneous group includes tibial metacarpal type [Rittler et al., 1990], mesomelic dysplasia [Burck, 1982], and humeral-metacarpal type, reported by Borochowitz [1991]. Our patient is clinically very similar to the Borochowitz [1991] patient, a healthy, intellectually normal 2½-year-old white girl with normal vision, rhizomelic shortness of humeri, punctate calcifications similar to our patient, and short hands.

Although Poznanski's [1994] subtypes of CDP-metacarpal type are designated for the specific long bone most strikingly involved, as a group these children demonstrate short stature, good general health, no skin lesions or cataracts, and short metacarpals as a common feature. This report of a second patient with CDP-humero-metacarpal type defines this as a separate nosological entity.

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